



HPRT1 gene

hypoxanthine phosphoribosyltransferase 1

Normal Function

The *HPRT1* gene provides instructions for producing an enzyme called hypoxanthine phosphoribosyltransferase 1. This enzyme allows cells to recycle purines, a type of building block of DNA and its chemical cousin RNA. Manufacturing purines uses more energy and takes more time than recycling purines, which makes recycling these molecules more efficient. Recycling purines ensures that cells have a plentiful supply of building blocks for the production of DNA and RNA. The process of recycling purines is also known as the purine salvage pathway.

Health Conditions Related to Genetic Changes

Lesch-Nyhan syndrome

More than 200 mutations in the *HPRT1* gene have been found to cause Lesch-Nyhan syndrome. These mutations include changes in single DNA building blocks (nucleotides) or insertions or deletions of small amounts of DNA within the gene. These changes result in either nonfunctional or very low-function hypoxanthine phosphoribosyltransferase 1. Under these conditions, uric acid, a waste product of purine breakdown, accumulates in the body and can cause gouty arthritis (arthritis caused by uric acid in the joints), kidney stones, and bladder stones. It is unclear how this enzyme deficiency causes the neurological and behavioral problems characteristic of Lesch-Nyhan syndrome.

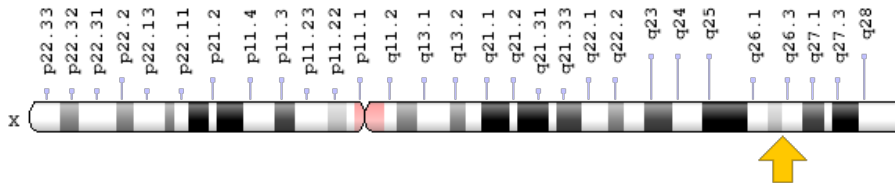
other disorders

Certain mutations in the *HPRT1* gene can also cause a condition featuring gouty arthritis called HPRT-related gout, previously known as Kelley-Seegmiller syndrome. Individuals with this condition have lower than normal levels of hypoxanthine phosphoribosyltransferase 1. Kidney problems commonly occur in people with this condition because a buildup of uric acid crystals can form kidney stones. Rarely, this condition will cause problems with the nervous system.

Chromosomal Location

Cytogenetic Location: Xq26.2-q26.3, which is the long (q) arm of the X chromosome between positions 26.2 and 26.3

Molecular Location: base pairs 134,460,145 to 134,500,668 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Guanine Phosphoribosyltransferase
- HGPRT
- HGPRTase
- HOX5.4
- HPRT
- HPRT_HUMAN
- HPRTase
- Hypoxanthine-Guanine Phosphoribosyltransferase
- hypoxanthine phosphoribosyltransferase 1 (Lesch-Nyhan syndrome)
- IMP Pyrophosphorylase

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002) Purine Bases Can Be Synthesized de Novo or Recycled by Salvage Pathways
<https://www.ncbi.nlm.nih.gov/books/NBK22385/>

GeneReviews

- Lesch-Nyhan Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1149>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28HPRT1%5BTI%5D%29+OR+%28%28Guanine+Phosphoribosyltransferase%5BTIAB%5D%29+OR+%28HGPRT%5BTIAB%5D%29+OR+%28hypoxanthine+phosphoribosyltransferase+1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

OMIM

- HYPOXANTHINE GUANINE PHOSPHORIBOSYLTRANSFERASE 1
<http://omim.org/entry/308000>
- KELLEY-SEEGMILLER SYNDROME
<http://omim.org/entry/300323>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_HPRT1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=HPRT1%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=5157
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3251>
- UniProt
<http://www.uniprot.org/uniprot/P00492>

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Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/HPRT1>

Reviewed: December 2007
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services